Locus 221 (rs1710354)

SNiPAcard

Block annotations

Block info

genomic range	chr19:51,802,140-51,802,650
block size	511 bp
variant count	2 variants

Basic features

Conservation/deleteriousness		Linked genes	
phyloP	μ = -1.125 [-1.962 – -0.287]	**gene(s) hit or close-by**	–
phastCons	μ = 0.003 [0 – 0.005]	**eQTL gene(s)**	CD33, ETFB
GERP++	μ = -0.726 [-1.13 – -0.323]	**potentially regulated gene(s)**	–
CADD score	μ = 1.387 [1.232 – 1.542]	**disease gene(s)**	ETFB

Trait annotations

Variant association

trait	min(p-value)	source DB	source entry/link	variant(s)
oleate (18:1n9)	3.00×10^{-5}	Metabolomics GWAS Server	24816252	2

Disease gene annotation

gene	trait	source DB	source entry/link	
ETFB	MULTIPLE ACYL-CoA DEHYDROGENASE DEFICIENCY	OMIM	MIM:231680	OMIM
ETFB	glutaric aciduria type 2B	DECIPHER	MIM:231680	OMIM
ETFB	Multiple acyl-CoA dehydrogenation deficiency, mild type	OrphaNet	OrphaNet:394532	orphanet
ETFB	Multiple acyl-CoA dehydrogenation deficiency, severe neonatal type	OrphaNet	OrphaNet:394529	orphanet

Direct effect on regulation

cis-eQTL

gene	transcript	probe	tissue	min(statistic) (type)	source	variant(s)
CD33	ENST00000262262	ILMN_1747622	blood	6.28×10^{-7} (p-value)	Westra et al.	1
CD33	ENST00000600557					
CD33	ENST00000601785					
CD33	ENST00000421133					
CD33	ENST00000598473					
ETFB	ENST00000596253	ILMN_1729374	blood	8.37×10^{-5} (p-value)	Westra et al.	2
ETFB	ENST00000593992					
ETFB	ENST00000309244					